

AMENDMENTS TO THE CLAIMS

1. (Currently Amended) A method of diagnosing myelinopathy in an individual, said myelinopathy resulting from a periaxin alteration in the individual, comprising the steps of:

obtaining a sample containing nucleic acid from said individual;

assaying said sample for an alteration in a periaxin polynucleotide, wherein said assaying step provides said diagnosis.

2. (Previously Presented) The method of claim 1, wherein said periaxin polynucleotide is SEQ ID NO:76.

3. (Original) The method of claim 1, wherein said periaxin polynucleotide is SEQ ID NO:1, SEQ ID NO:27, SEQ ID NO:28, SEQ ID NO:29, SEQ ID NO:30, SEQ ID NO:31, SEQ ID NO:32, SEQ ID NO:33, SEQ ID NO:34, SEQ ID NO:35, SEQ ID NO:36, SEQ ID NO:37, SEQ ID NO:38, SEQ ID NO:39, SEQ ID NO:40, SEQ ID NO:41, SEQ ID NO:42, SEQ ID NO:43, SEQ ID NO:44, SEQ ID NO:45, SEQ ID NO:46, SEQ ID NO:47, SEQ ID NO:48, SEQ ID NO:49, SEQ ID NO:50, SEQ ID NO:51, SEQ ID NO:52, SEQ ID NO:53, SEQ ID NO:54, SEQ ID NO:55, SEQ ID NO:56, SEQ ID NO:57, SEQ ID NO:58, SEQ ID NO:59, SEQ ID NO:60, SEQ ID NO:61, SEQ ID NO:62, SEQ ID NO:63, SEQ ID NO:64, SEQ ID NO:65, SEQ ID NO:66, SEQ ID NO:67, SEQ ID NO:68, SEQ ID NO:69, SEQ ID NO:70, SEQ ID NO:71, SEQ ID NO:72, SEQ ID NO:73, SEQ ID NO:74, SEQ ID NO:75, SEQ ID NO:76, or SEQ ID NO:77.

4. (Currently Amended) The method of claim 1, wherein said myelinopathy is selected from the group consisting of Charcot-Marie-Tooth (CMT) syndrome, hereditary neuropathy with liability to pressure palsies (HNPP), Dejerine-Sottas syndrome (~~DSSDSN~~), congenital hypomyelinating neuropathy (CHN), and Roussy-Levy syndrome (RLS).

5. (Original) The method of claim 1, wherein said assaying step further comprises a polymerase chain reaction.

6. (Original) The method of claim 5, wherein primers for said polymerase chain reaction are selected from the group consisting of SEQ ID NO:3, SEQ ID NO:4, SEQ ID NO:5, SEQ ID NO:6, SEQ ID NO:7, SEQ ID NO:8, SEQ ID NO:9, SEQ ID NO:10, SEQ ID 25435403.1

NO:11, SEQ ID NO:12, SEQ ID NO:13, SEQ ID NO:14, SEQ ID NO:15, SEQ ID NO:16, SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20, SEQ ID NO:21, SEQ ID NO:22, SEQ ID NO:23, SEQ ID NO:24, SEQ ID NO:25, and SEQ ID NO:26.

7. (Previously Presented) The method of claim 1, wherein said alteration is 3775G>A, 1216G>A, 4075-4077d, 1483G>C, 3394A>G, 3248C>G, 2763A>G, 2645C>T, 306C>T, 1491C>G, 2655T>C, 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.

Claims 8-34. (Cancelled)

35. (Currently Amended) A method of detecting the presence or absence of a mutation associated with a myelinopathy, said myelinopathy resulting from a periaxin mutation in the individual, the method comprising:

- a) isolating a test nucleic acid from a subject, said test nucleic acid comprising a periaxin polynucleotide;
- b) comparing the test nucleic acid to a reference wild-type periaxin polynucleotide; and
- c) determining the differences between the test nucleic acid and the reference wild-type periaxin polynucleotide, wherein the differences are mutations in the periaxin polynucleotide of the subject, and wherein said detection of the presence or absence of the mutation is therein provided.

36. (Previously Presented) The method of claim 35, wherein said mutation is 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.

37. (Previously Presented) The method of claim 35, wherein said mutation encodes a defect of a periaxin polypeptide, wherein the defect is R953X, R368X, S929fsX957, R196X, V763fsX774, C715X, or R82fsX96.

38. (Original) The method of claim 35, wherein said periaxin polynucleotide is SEQ ID NO:1, SEQ ID NO:27, SEQ ID NO:28, SEQ ID NO:29, SEQ ID NO:30, SEQ ID NO:31, SEQ ID NO:32, SEQ ID NO:33, SEQ ID NO:34, SEQ ID NO:35, SEQ ID NO:36, SEQ ID NO:37, SEQ ID NO:38, SEQ ID NO:39, SEQ ID NO:40, SEQ ID NO:41, SEQ ID NO:42, SEQ ID NO:43, SEQ ID NO:44, SEQ ID NO:45, SEQ ID NO:46, SEQ ID NO:47,

SEQ ID NO:48, SEQ ID NO:49, SEQ ID NO:50, SEQ ID NO:51, SEQ ID NO:52, SEQ ID NO:53, SEQ ID NO:54, SEQ ID NO:55, SEQ ID NO:56, SEQ ID NO:57, SEQ ID NO:58, SEQ ID NO:59, SEQ ID NO:60, SEQ ID NO:61, SEQ ID NO:62, SEQ ID NO:63, SEQ ID NO:64, SEQ ID NO:65, SEQ ID NO:66, SEQ ID NO:67, SEQ ID NO:68, SEQ ID NO:69, SEQ ID NO:70, SEQ ID NO:71, SEQ ID NO:72, SEQ ID NO:73, SEQ ID NO:74, SEQ ID NO:75, SEQ ID NO:76, or SEQ ID NO:77.

39. (Original) The method of claim 35, wherein said comparing step is by DHPLC, sequencing, hybridization, or a combination thereof.

40. (Currently Amended) The method of claim 35, wherein the myelinopathy is Charcot-Marie-Tooth (CMT) syndrome, hereditary neuropathy with liability to pressure palsies (HNPP), Dejerine-Sottas syndrome (~~DSSDSN~~), congenital hypomyelinating neuropathy (CHN), or Roussy-Levy Syndrome (RLS).

41. (Cancelled).

42. (Previously Presented) The method of claim 35, wherein said mutation encodes a defect of a periaxin polypeptide, wherein the defect is E1259K, A406T, E1359delΔ, E495Q, R1132G, P1083R, I921M, A882V, T102T, P497P, or P885P.

43. ((Currently Amended)) A method of diagnosing myelinopathy in an individual comprising the steps of:

obtaining a sample containing nucleic acid from said individual; assaying said sample for an alteration in a periaxin polynucleotide, wherein said alteration is associated with said myelinopathy, and wherein said myelinopathy comprises a prominent sensory neuropathy, wherein said assay provides said diagnosis.

44. (Previously Presented) The method of claim 43, wherein said periaxin polynucleotide is SEQ ID NO:76.

45. (Previously Presented) The method of claim 43, wherein said periaxin polynucleotide is SEQ ID NO:1, SEQ ID NO:27, SEQ ID NO:28, SEQ ID NO:29, SEQ ID NO:30, SEQ ID NO:31, SEQ ID

NO:32, SEQ ID NO:33, SEQ ID NO:34, SEQ ID NO:35, SEQ ID NO:36, SEQ ID NO:37, SEQ ID NO:38, SEQ ID NO:39, SEQ ID NO:40, SEQ ID NO:41, SEQ ID NO:42, SEQ ID NO:43, SEQ ID NO:44, SEQ ID NO:45, SEQ ID NO:46, SEQ ID NO:47, SEQ ID NO:48, SEQ ID NO:49, SEQ ID NO:50, SEQ ID NO:51, SEQ ID NO:52, SEQ ID NO:53, SEQ ID NO:54, SEQ ID NO:55, SEQ ID NO:56, SEQ ID NO:57, SEQ ID NO:58, SEQ ID NO:59, SEQ ID NO:60, SEQ ID NO:61, SEQ ID NO:62, SEQ ID NO:63, SEQ ID NO:64, SEQ ID NO:65, SEQ ID NO:66, SEQ ID NO:67, SEQ ID NO:68, SEQ ID NO:69, SEQ ID NO:70, SEQ ID NO:71, SEQ ID NO:72, SEQ ID NO:73, SEQ ID NO:74, SEQ ID NO:75, SEQ ID NO:76, or SEQ ID NO:77.

46. (Previously Presented) The method of claim 43, wherein said assaying step further comprises a polymerase chain reaction.
47. (Previously Presented) The method of claim 46, wherein primers for said polymerase chain reaction are selected from the group consisting of SEQ ID NO:3, SEQ ID NO:4, SEQ ID NO:5, SEQ ID NO:6, SEQ ID NO:7, SEQ ID NO:8, SEQ ID NO:9, SEQ ID NO:10, SEQ ID NO:11, SEQ ID NO:12, SEQ ID NO:13, SEQ ID NO:14, SEQ ID NO:15, SEQ ID NO:16, SEQ ID NO:17, SEQ ID NO:18, SEQ ID NO:19, SEQ ID NO:20, SEQ ID NO:21, SEQ ID NO:22, SEQ ID NO:23, SEQ ID NO:24, SEQ ID NO:25, and SEQ ID NO:26.
48. (Previously Presented) The method of claim 43, wherein said alteration is 3775G>A, 1216G>A, 4075-4077d, 1483G>C, 3394A>G, 3248C>G, 2763A>G, 2645C>T, 306C>T, 1491C>G, 2655T>C, 2145T>A, 1102C>T, 2289delT, 2787delC, 2857C>T, or 247ΔC.
49. (Previously Presented) A method of detecting a polymorphism or mutation in a periaxin polynucleotide of an individual, comprising the steps of:

obtaining a sample comprising said periaxin polynucleotide from said individual; assaying said periaxin polynucleotide for the polymorphism or mutation.

50. (Previously Presented) The method of claim 49, wherein said periaxin polynucleotide comprises SEQ ID NO:76.

51. (New) The method of claim 1, wherein said myelinopathy is Dejerine-Sottas syndrome.

52. (New) The method of claim 1, wherein said individual is suspected of having the myelinopathy.

53. (New) The method of claim 1, wherein the alteration comprises a homozygous periaxin mutation.

54. (New) The method of claim 1, wherein the alteration comprises a compound heterozygous periaxin mutation.

55. (New) The method of claim 43, wherein the alteration comprises a homozygous periaxin mutation.

56. (New) The method of claim 43, wherein the alteration comprises a compound heterozygous periaxin mutation.

57. (New) A method of identifying an individual suspected of having myelinopathy or being a carrier of myelinopathy, comprising the steps of:

obtaining from said individual a sample comprising nucleic acid; and assaying said sample for an alteration in a periaxin polynucleotide, wherein the presence of the alteration identifies said individual as having periaxin-associated myelinopathy or being a carrier of periaxin-associated myelinopathy.

58. (New) The method of claim 57, wherein said myelinopathy comprises a prominent sensory neuropathy.

59. (New) The method of claim 57, wherein the alteration comprises a homozygous periaxin mutation.

60. (New) The method of claim 57, wherein the alteration comprises a compound heterozygous periaxin mutation.

61. (New) A method of identifying an individual suspected of having myelinopathy or being a carrier of myelinopathy, comprising the steps of:

obtaining from said individual a sample comprising genomic DNA having two PRX alleles; and

assaying said sample for an alteration in a periaxin polynucleotide, wherein the presence of the alteration in the periaxin polynucleotide is indicative of an alteration in at least one of the PRX alleles, wherein the presence of the alteration in both PRX alleles identifies said individual as having periaxin-associated myelinopathy and wherein the presence of the alteration in one allele identifies said individual as being a carrier of periaxin-associated myelinopathy.